

SOUTHWESTERN NEWS

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FLY GENE MAY BE BASIS FOR INFERTILITY TESTING, TREATMENT

DALLAS — June 27, 1996 — Could male sterility be in the genes? Researchers at UT Southwestern Medical Center at Dallas think so. They say a mutated gene in the fruit fly (*Drosophila*) may give hope to the 2 percent of men who are infertile because they don't make enough sperm.

Writing in the June 27 issue of the journal *Nature*, Dr. Steven A. Wasserman, associate professor of biochemistry, and his colleagues, biochemistry fellow Dr. Jean Z. Maines and predoctoral fellow Charles G. Eberhart, report cloning a fly version of the human gene *Deleted in Azoospermia*, or *DAZ*. Men lacking azoospermia factor produce semen without sperm. Many of those men — at least one in 8,000 — have defects in a section of the Y chromosome where the *DAZ* gene resides.

"Studies on human azoospermia factor have identified for the first time a gene whose sole function appears to be in human male fertility," Wasserman said.

The fly version of the *DAZ* gene, named *boule*, was first identified in 1993 by Wasserman. He said that flies lacking *boule* make no sperm and that the *boule* gene expresses a similar protein to the one expressed by *DAZ*. These common factors "provide strong evidence that *DAZ* is the azoospermia factor, and that *boule* is the counterpart of *DAZ*."

Wasserman explained that mutations in *boule* and *DAZ* block sperm production at the

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same stage — just before the chromosomes are partitioned among the developing sperm.

Once chromosomes are partitioned, the sperm mature and are ready to fertilize eggs. Like defects in the azoospermia factor, mutations in *boule* appear to only affect males, and both *boule* and *DAZ* are only found in the testes.

Wasserman's research has significant implications for many men. In addition to those with azoospermia, a large number with low sperm count, or oligospermia, also have a defective *DAZ* gene. "My own guess is that one in a thousand, or one in 2,000 men are likely to have a mutation," he said.

These findings also raise the possibility of gene therapy for treating male infertility. "The better we can understand the gene at the molecular level, the better chance we have of being able to design a treatment for infertility," Wasserman said.

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